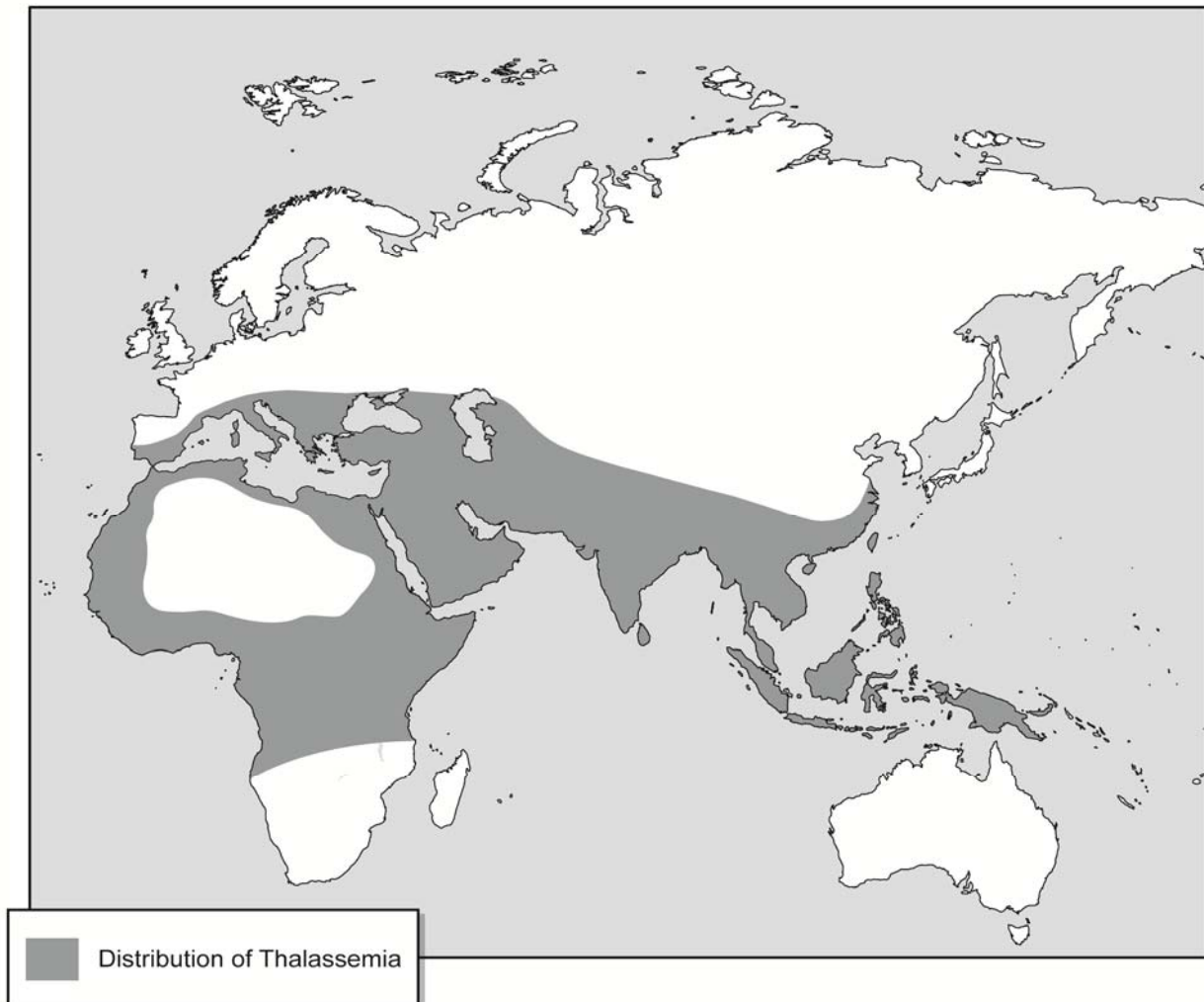


The Epidemiology of Thalassemia

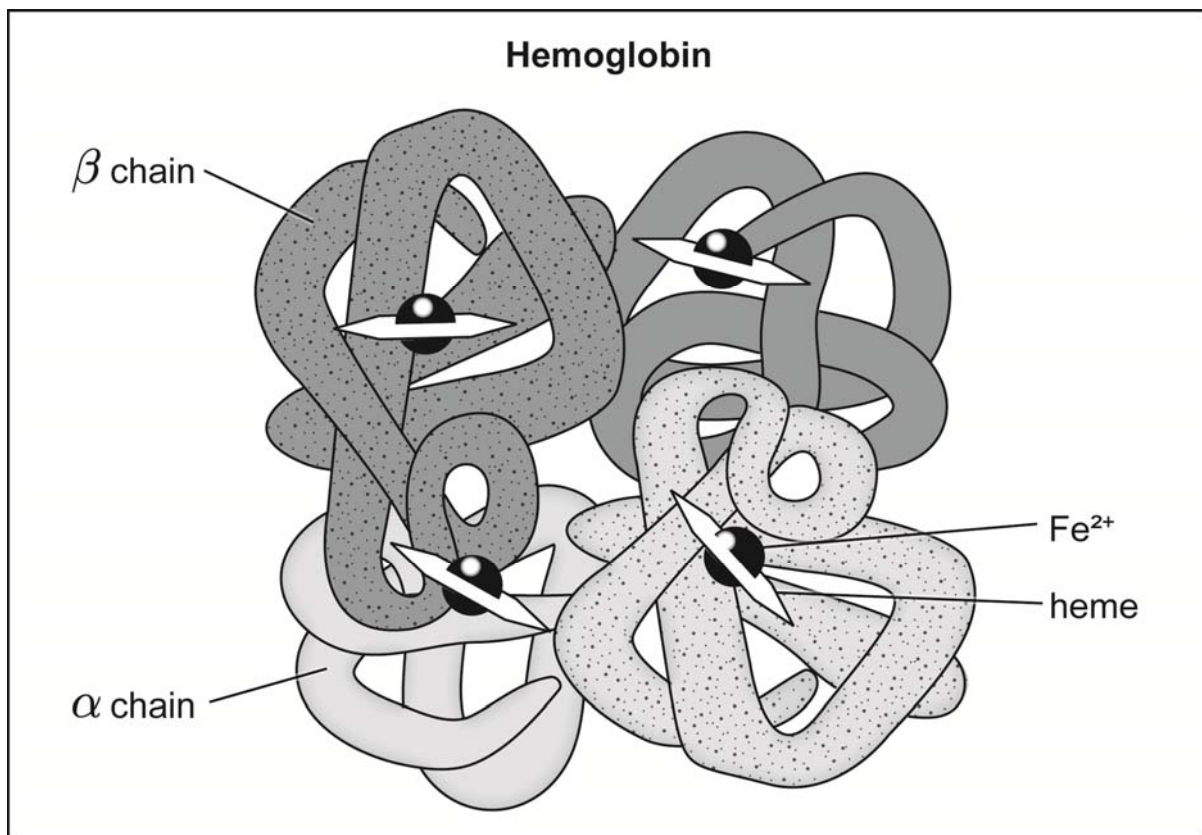


The Alpha-Globin Gene and Alpha-Thalassemia

Part 1

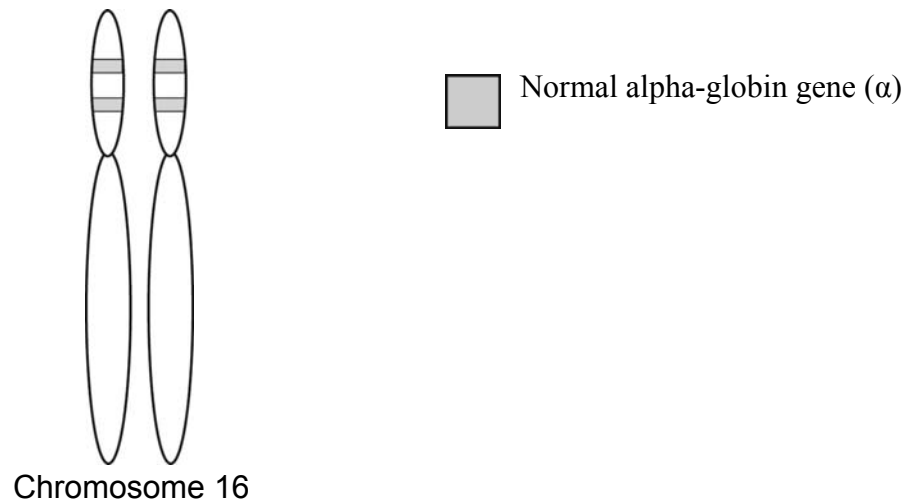
Alpha-thalassemia is caused by problems in the production of the alpha globin protein in hemoglobin.

Hemoglobin is normally made up of two alpha globin protein chains and two beta globin protein chains. Different genes code for the alpha and beta protein chains.

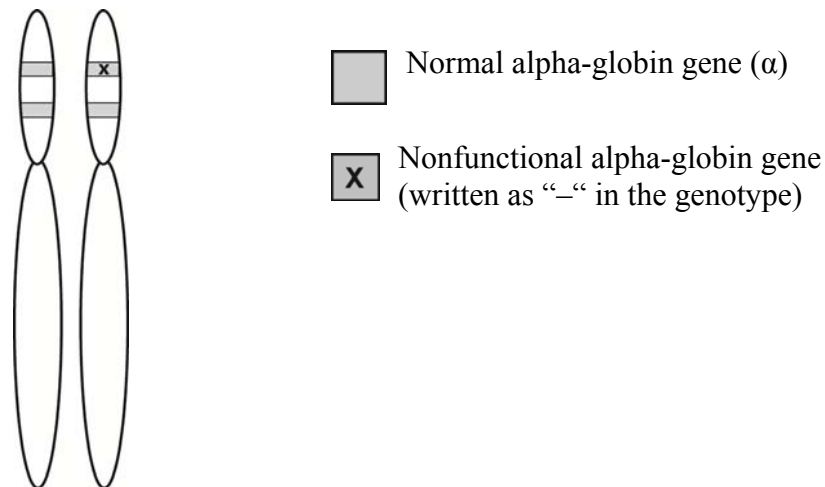


Alpha-Globin Gene

The alpha globin protein is coded for by the alpha-globin gene which is on chromosome 16 in humans. Humans normally have four copies of the alpha-globin gene.



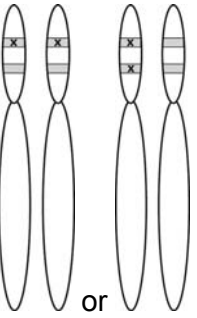


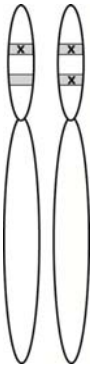
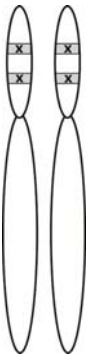
Individuals with alpha-thalassemia have a problem with one or more of their alpha-globin genes.



Individuals with alpha-thalassemia do not make as much alpha-globin protein as normal individuals do. The amount of protein made depends on how many working copies of the alpha-globin gene that the person has.

Part 2

Number of functional copies of the alpha-globin gene	Number of non-functional copies of the alpha-globin gene	Chromosome Representation	Name of disease	Symptoms
4	0		Normal condition (healthy individual)	
3	1		Silent carrier α^+ -thalassemia	<ul style="list-style-type: none"> • Usually no symptoms and no anemia • Blood tests usually normal • Hemoglobin normal • Slight changes in size of red blood cells (smaller than normal; microcytosis) • Slight hypochromia (light color of red blood cells)
2	2		Alpha-thalassemia trait or α^0 -thalassemia	<ul style="list-style-type: none"> • Mild anemia • Small red blood cells (microcytosis) • Hypochromia (light color of red blood cells) • Blood tests usually normal • Hemoglobin normal

1	3		Hemoglobin H (HbH) disease	<ul style="list-style-type: none"> • Moderate to severe anemia • Small (microcytic) red blood cells • Hypochromia (light, pale color of red blood cells) • Fatigue • Mild jaundice • Enlarged spleen • Bone deformities (in some cases)
0	4		Alpha-thalassemia major or Hemoglobin Bart hydrops fetalis (Hb Bart) syndrome	<ul style="list-style-type: none"> • Usually fatal before or shortly after birth